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PERIODICALS

AMERICAN JOURNAL OF HUMAN GENETICS
July, 1966, Vol. 18, No. 4.

Population Dynamics of Tay-Sachs Disease. 1. Reproductive Fitness and Selection *by* NTINOS C. MYRIANTHOPOULOS *and* STANLEY M. ARONSON.

Possible explanations for the higher frequency of Tay-Sachs Disease in Jewish than in non-Jewish populations are explored. Genetic drift, intermarriage between genetically distinct groups, different mutation rates in the two populations are all discounted. The high consanguinity rate among Sephardic Jews might have led to the elimination of the gene, resulting in the present low incidence of the condition in this group but such a mechanism could not account for the low frequency in non-Jewish peoples. Heterozygote advantage may be a factor in maintaining the gene frequency in certain Jewish groups, notably the Ashkenazi Jews, for the authors show that the grandparents of index patients are more fertile than those of a control group. A statistically significant difference is not shown but would not be expected in a small sample since there need only be a small increase in fertility to maintain the higher gene frequencies.

Some Further Observations on the Sex Ratio among Infants born to Survivors of the Atomic Bombings of Hiroshima and Nagasaki *by* WILLIAM J. SCHULL, JAMES V. NEEL *and* A. HASHIZUME.

Data gleaned from a further 47,624 births in these two Japanese cities now make it unlikely that exposure of one or both parents to the atomic bombings had any effect on the sex ratio. The authors attempt to explain this apparent reversal of conclusions drawn from earlier data.

A Cohort-Type Study of Survival in the Children of Parents exposed to Atomic Bombings *by* HIROO KATO, WILLIAM J. SCHULL *and* JAMES V. NEEL.

In 1956, the study made by Neel and Schull showed no evidence of an increased death rate among children of survivors of the atomic bombings, so far as the immediate neonatal period was concerned. Now, all children born between 1st May 1946 and 31st December 1958 to parents exposed to radiation have been followed, the average number of years "at risk" being nine. After due allowance for such factors as parental age, parity, etc., there was no indication that the exposure of parents to atomic bombs had altered the life expectancy of their children. From the data collected, a minimal doubling dose of radiation of the type emitted by an atomic bomb is estimated as 50 r, for mutations which lead to early death in the first post-radiated generation.

Frequency and Occurrence of Chromosomal Syndromes. I. D-Trisomy; II. E-Trisomy *by* PATRICK E. CONEN *and* BAYZAR ERKMAN.

D-Trisomy probably occurs in about one in 14,500 live births; E-Trisomy in about one in 4,500 live births. Of the D-trisomics, four out of nine had a D/D translocation: the only clinical difference found between trisomic and translocation cases was the greater severity of cardiovascular lesions in the former.

For babies with both types of chromosome aberration, such features as the sex ratio, survival period and maternal age are given for the present series and contrasted with those of cases previously published.

Some Serum Protein Polymorphisms in Kalahari Bushmen and Bantu: Gamma Globulins, Haptoglobins and Transferrins *by* TREFOR JENKINS *and* ARTHUR G. STEINBERG.

Bushmen of Kalahari have the lowest frequency for the Hp^1 gene found in any population indigenous to Africa. Inv. (1) gene frequencies for both Bushmen and Bantu are similar to West African and American Negroes. Gm factors show resemblances to both Negro and Mongoloid populations but Bushmen have three Gm alleles not found in other African peoples. These "unique" alleles will help in assessing the extent of Bushman admixture in Bantu tribes.

Frequency of Polymorphic Types of Red Cell Enzymes and Serum Factors in Alaskan Eskimos and Indians *by* EDWARD M. SCOTT, IRMA W. DUNCAN, VIRGINIA EKSTRAND *and* RITA C. WRIGHT.

The P^c gene or erythrocyte acid phosphatase was not found in Eskimos or Indians; there was a high frequency of P^a in all the groups studied. Phosphoglucomutase types were similar in Eskimos, Aleuts and Indians but population differences were revealed in the Hp and Gc systems.

HELEN BLYTH

ANNALS OF HUMAN GENETICS

November, 1965, Vol. 29, Part 2.

Racial Admixture in North-eastern Brazil *by* H. KRIEGER, N. E. MORTON, M.P. MI, ELIANE AZEVÊDO, A. FREIRE-MAIA *and* N. YASUDA.

Statistical examination of blood group data from a north-eastern Brazilian population indicates that the racial mixture is about 30 per cent Negro, 11 per cent Indian and 59 per cent Caucasian.

The Antimode and Lines of Optimal Separation in a Genetically Determined Bimodal Distribution, with Particular Reference to Phenylthiocarbamide Sensitivity *by* H. KALMUS *and* SHEILA MAYNARD SMITH.

It is suggested that it may be better to divide a bimodal population so that equal numbers, or equal proportions, of each group are misclassified rather than at the antimode. A method of doing this on the assumption of incomplete dominance is developed and is illustrated from PTC taste threshold data.

The Relative Positions of the Chromosomes in the Human Cell in Mitosis *by* D. E. BARTON, FLORENCE N. DAVID *and* MAXINE MERRINGTON.

Chromosomes 13–15 and 20–22 tend to lie near the centre and chromosome 6 and the sex chromosomes near the periphery of the nucleus.

Cytological Studies on a Human Ring Chromosome *by* PATRICIA COOKE *and* R. R. GORDON.

Chromosome studies are reported in a case of microcephalic dwarfism with a ring chromosome. It is concluded that the ring was formed from the terminal deletion of about 10 per cent of one of the number 1 chromosomes.

Haemoglobin E and β-thalassaemia: their Distribution in Thailand *by* G. FLATZ, C. PIK *and* S. SRINGAM.

The frequencies of Haemoglobin E and β-thalassaemia in different geographical and ethnic groups in Thailand are reported and discussed.

Sex-linked Recessive Congenital Deafness and the Excess of Males in Profound Childhood Deafness *by* G. R. FRASER.

Familial data on congenital deafness are presented and it is concluded that about 3 per cent of all the hereditary cases are due to a sex-linked recessive gene. The nature of the defect in these cases is discussed.

The Nail-patella Syndrome. A study of a Caernarvon family *by* C. A. CLARKE *and* E. WYN JONES.

A pedigree is reported in which the genes controlling the nail-patella syndrome and the A_1 blood group are linked in coupling.

A Child with an Extra Small Metacentric Chromosome *by* M. D'A CRAWFURD *and* KUSUM P. LELE.

A case is reported in which there were five small metacentric and three small acrocentric chromosomes. The origin of the anomaly is discussed.

On the Geometry of Loops and Deltas *by* L. S. PENROSE.

The geometry of digital ridges is discussed.

M. G. BULMER

JOURNAL OF MEDICAL GENETICS
September, 1966, Vol. 3, No. 3.

Expectation of Abnormality on Paternal and Maternal Sides: A Computational Model *by* ELIOT SLATER.

It is difficult to distinguish the effects of inheritance by a dominant major gene with diminished penetrance from polygenic inheritance. Nevertheless, it seems to be worth while making the distinction in the case of genetically determined abnormalities, if this is practicable. The possibility has been considered that, if families are sought for in which relatives on the paternal or maternal side include two or more secondary cases of the condition under investigation, single gene inheritance might be exhibited in a preponderance of cases on either the paternal or maternal side, while polygenic inheritance might tend to go with a more even distribution of secondary cases between the two sides of the ascendance. A computational model, making use of some simplifying hypotheses, has been set up to test this possibility. It is found that, in the cases of near relatives such as parents or sibs of parents, even with polygenic inheritance, one expects a unilateral preponderance of secondary cases, rather than a more even distribution. This does not exclude the possibility of distinguishing between the consequences of the two alternative hypotheses, if reliable accounts of more distant relatives are available, or if use can be made of families with three or more secondary cases.

The X-linked Blood Group System Xg: Tests on British, Northern American and Northern European unrelated people and families *by* J. NOADES, J. GAVIN, P. TIPPETT, R. SANGER *and* R. R. RACE.

The Xg groups are recorded of 3,418 samples from unrelated white people in Britain, North America, and from the mainland of Northern Europe. The calculated gene frequencies are:

	Great Britain	N. America	N. Europe
Xg^a	0·675	0·671	0·600
Xg	0·325	0·329	0·400

These frequencies are used to analyse the results of testing 1,339 families with 3,113 children: four families include apparent exceptions to the rules of X-linked inheritance.

Gene Effect in Carriers of Anhidrotic Ectodermal Dysplasia *by* C. B. KERR, R. S. WELLS *and* K. E. COOPER.

Examination of females heterozygous for anhidrotic ectodermal dysplasia revealed partial anodontia. Results of thermally-induced sweating and histological examination of skin sections indicated a quantitative deficiency of sweat gland as compared with controls. The findings were consistant with the inactive X-chromosome theory.

Haemoglobin Genotypes, ABO Blood Groups and Burkitt's Tumour *by* A. OLUFEMI WILLIAMS.

In a series of 100 Yoruba children over the age of five years with clinico-pathological evidence of malignant lymphoma, there is evidence that children with the AA genotype on haemoglobin electrophoresis are more susceptible to development of the tumour. There is no significant difference between the blood group distribution of patients and controls.

Blood Group Changes in Leukaemia *by* M. AYRES, F. M. SALZANO *and* O. K. LUDWIG.

Fifty-one patients with leukaemia, living in the State of Rio Grande, were tested in relation to ABO, MN and Rh blood groups. A search was also made for changes their in χ and β agglutinins. The I antigen was studied in the blood of twenty-five patients. In twenty-eight patients saliva was tested for the secretion of ABH and Le^a substances.

Nine patients showed antigenic abnormalities in their blood, the more common being related to the H and I antigens. The changes do not seem to be more prevalent in any type of leukaemia. No abnormalities were detected in the saliva and plasma studies.

Satellite Association and Translocation Mongolism *by* H. ZELLWEGER, G. A. ABBO *and* R. CUANY.

Four translocation mongols, three sporadic and one familial, and their families were examined for association of satellited chromosomes. Evidence of increased satellite association was found in

three mongoloids and in one of their respective parents. The other parent of these three mongoloids and the fourth translocation mongoloid and both of his parents had normal SA. The data suggest that increased SA may be one, but not the only, cause of translocation mongolism, and that the tendency towards increased SA may be transmitted as a familial, perhaps inherited, trait.

The Silent Gene for Serum Pseudocholinesterase *by* A. SZEINBERG, S. PIPANO, E. OSTFELD *and* L. EVIATAR.

A family showing complete absence of serum pseudocholinesterase activity in father and two sons is described. The family data are compatible with the hypothesis that these cases were homozygous for the "silent" gene allelic to the "normal" gene.

Genetic considerations from this and previously reported families exclude an alternative hypothesis, that the "silent" gene is independent of the E_1 locus and suppresses the action of the E_1^a gene, with a probability of about 0·00003.

Homocystinuria: An Observation on the Inheritance of Cystathionine Synthase Deficiency *by* G. GAULL *and* M. K. GAITONDE.

Cystathionine synthase activity, previously shown to be absent from liver and brain of patients with homocystinuria, was found to be present in the optic lens of such a patient. The biological implications are briefly discussed.

Genetic Diversity in Serum Albumin *by* MORTON S. ADAMS.

A genetic and biochemical study is presented of a slowly migrating electrophoretic species of human serum albumin occurring in twenty-two members of the family of a seven-year-old boy with the nephrotic syndrome. This trait is controlled by a rare co-dominant allele of normal albumin. The published reports are reviewed and the significance of the genetic diversity of serum albumin is discussed.

The Use of the Surname as a Genetic Marker in Wales *by* D. J. B. ASHLEY *and* H. D. DAVIES.

Evidence is adduced to support the hypothesis that the surname can be used as a marker for Welshness in the heterogenous population of Wales, and that this marker may be used in the study of the genetic components of illness, immunities and other traits. It is thought that the technique will not be valid in one or two generations hence.

A Monopodal Sireniform Monster with Dermatoglyphic and Cytogenetic Studies *by* M. D'A. CRAWFURD, S. R. ISMAIL *and* J. S. WIGGLESWORTH.

A case is reported of a monopodal sireniform stillborn male foetus. The child was one o twins, the other twin being a liveborn normal male. Nuclear sex and chromosome constitution were of the normal male pattern. The features and possible aetiology of sympodia and monopodia are discussed.

A Pair of Twins, One of Whom has Chronic Granulocytic Leukaemia *by* L. DOUGAN, IAN D. SCOTT *and* H. J. WOODLIFFE.

A pair of monozygous twins, one of whom had chronic granulocytic leukaemia is described. No apparent environmental stimulus to leukaemogenesis was found.

Turner's Syndrome in a Phenotypic Male with XO/XY Mosaicism and Autosomal Aberrations *by* M. C. URMENYI, M. K. BEATTIE *and* M. R. MIRZA.

An elderly phenotypic male is described with Turner's syndrome, anorchia, congenital cysts of the kidney, mental subnormality with schizophrenic overlay, XO-XY karyotype, and aberrations of chromosomes of the C and E groups. Examination of a limited number of metaphases cultured from the mother's blood suggested the possibility of maternal transmission.

Apparent Deletion of X Chromosome in a Prepuberal Girl *by* E. STEINBERGER, A. STEINBERGER, K. D. SMITH *and* W. H. PERLOFF.

Chromosome studies on an eight-year-old girl with short stature, high arched palate and loss of eyebrows revealed 46 chromosomes, one normal X chromosome and an extra chromosome

morphologically similar to the chromosomes of the D group. Autoradiographic studies revealed consistent late labelling of the abnormal long acrocentric chromosomes.

A Proposed Classification of Genetically Determined Mosaicism in Man *by* M. J. COREY *and* J. R. MILLER.

The term "mosaicism" is now used extensively in human cytogenetics. In fact, the term has had long usage and specific meanings in human genetics. A proposed classification is presented.

D. J. MANTLE

POPULATION STUDIES

November 1966, Vol. 20, No. 2.

The Decline of Fertility: Innovation or Adjustment Process *by* G. CARLSSON.

After studying the level of fertility at different times, and examining both the pace of change over the past hundred years and the rate of its diffusion both socially and geographically, the author concludes that the big decline in family size in Western Europe was the consequence of a change in aims and ideals rather than the adoption of family planning for the first time.

Malthus on Norway *by* M. DRAKE.

Much of the *Essay on the Principle of Population* is based on Malthus's Scandinavian tour The author here shows that Malthus's use of non-statistical sources, and the bias of his itinerary and informants, led him into errors, particularly in under-estimating mortality. As a consequence, the basis of his theory is to some extent undermined.

Economic Considerations in Family Growth Decisions *by* R. FREEDMAN *and* L. COOMBS.

Studies of data collected in Detroit suggest that the timing of pregnancies is more closely related to family income than is the total number of children borne. Possibly a more important factor than the actual level of income is the potential parents' evaluation of their economic position, in relation to their needs in other respects and in relation to the wealth of their neighbours.

Parameters of the Menstrual Cycle: A Reply by R. G. POTTER.

This is a rejoinder to a contribution by W. H. James in the July 1965 issue of *Population Studies*. James defended the menstrual statistics of E. J. Farris, but Potter argues that they are open to the suspicion of bias. He thinks, however, that the methods of James should prove useful in the future.

Bridal Pregnancy in Rural England in Earlier Centuries *by* P. E. H. HAIR.

A special analysis of parish registers in a number of villages over three centuries shows that at least one-sixth and probably one-third of marriages were followed by a maternity within nine months. The propensity to be pregnant at marriage was higher in the later part of the period than it had been in the earlier part.

On the Probable Age Structure of the Roman Population *by* K. HOPKINS.

The distribution of ages at death shown on Roman tombstones gives rise to improbable mortality figures, judged in relation to the UN model life tables, and also varies appreciably with geographical region. Distribution is attributable to the customs of the period, e.g. a wife is more likely to be commemorated if she pre-deceased her husband than if she survived him. The author concludes that these records are of no value as evidence for estimating the expectation of life.

P. R. C.

Contents

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